Running head: DETRIMENTAL EFFECTS

Lack of Awareness of Hemochromatosis in the Healthcare Community and the Detrimental Effects of Late Diagnosis

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I. Introduction and Literature Review

Hereditary hemochromatosis (HH) is an autosomal recessive blood disorder that is characterized by an increase in iron absorption, which, if untreated, can lead to excessive iron deposits in organs and eventual organ failure (Phatak, Bonkovsky, & Kowdley, 2008, p. 270). HH is caused by a mutation of the HFE gene that regulates iron absorption. Since the disease is recessive, a person must have two mutated genes in order for the disorder to be expressed (termed homozygous). The two main mutations that can occur are C282Y mutation (accounting for 90-95% of cases) and H63D mutation. H63D is associated with a less severe form of iron absorption than a C282Y mutation. Although fairly uncommon, people can also exhibit signs and symptoms of HH if they possess only one mutated gene (termed heterozygous), but generally they are only carriers in this case (Plaut & McLellan, 2009, p. 18).

Most people absorb roughly one milligram of iron per day from their diets, which is then equally excreted through sweat, sloughing skin cells, urine, and the gastrointestinal tract. In contrast, people with HH can absorb anywhere from two to four milligrams per day, but the excretion rate remains the same (Schrier & Bacon, 2008, p. 2). Premenopausal women tend to have lower iron levels and fewer complications from HH than men since they lose blood (and iron) during menstruation (Bring, Partovi, Ford, &Yoshida, 2008, p. 333). Over time the body begins to deposit excess iron in organs such as the liver, heart, brain, pancreas, and lungs (Plaut & McLellen, 2009, p. 18). Common symptoms of HH include extreme fatigue, impotence and infertility, skin hyperpigmentation (also referred to as bronzing), depression, abdominal pain, arthritis/joint pain, diabetes, liver disease, and heart disease (Weinberg, 2004, p. 9). Diagnosis typically does not occur until between the ages 40 to 60, when iron levels have already reached toxic, damaging levels (Plaut & McLellen, 2009, p. 18).

Although once thought to be a rare disorder, recent research has shown that HH is much more common than originally believed. Approximately ten percent of the Caucasian population in the United States and Western Europe are heterozygous, thus, carriers of the disease. About one in every two-hundred people (0.5 percent) is homozygous, therefore, will show clinical manifestations of the disease (Schrier & Bacon, 2008, p. 1). Despite the frequent incidence of HH, the disorder often goes undiagnosed and untreated until the late stages of the disease. In one study, patients reported that before receiving the diagnosis of HH, they saw an average of three different doctors and the correct diagnosis took more than nine years. Common misdiagnoses include arthritis, gallbladder and liver disease, stomach disorders, diabetes, mental health disorders, and hormonal deficiencies. In many cases these diagnoses were indeed correct, but did not identify the underlying cause of the problem: iron overload (Weinberg, 2004, p.9). Failure to correctly identify HH is particularly problematic, because another study showed that people who receive early diagnosis and treatment have an improved life expectancy of seven years (Phatak et al., 2008, p. 271).

Since the disease is common and complications can be easily prevented with early diagnosis, the question of community screening has been raised. Surprisingly, much debate has ensued. Concerns—at both national and international levels—include: incomplete knowledge "about disease penetrance, the potential for discrimination in insurance and employment, whether anxiety would be caused by screening of individuals unaware of their genetic status, and whether screening should be by Fe [iron] studies or genetic testing" (Allen, Nisselle, Collins, Williamson, & Delatycki, 2008, p. 363).

II. Research Question/ Hypothesis

Is lack of awareness about the existence and prevalence of hereditary hemochromatosis among medical personnel responsible for late diagnosis of the disease, resulting in improper treatment, increased medical expenses, and life-threatening complications?

III. Methodology

The majority of the research for my thesis will be based on the use of preexisting literature and scientific studies that have taken place within the last five years. I will review books and scholarly journals regarding iron, iron overload, hereditary hemochromatosis, and iron disorders. In order to evaluate whether or not the literature is reliable, I will assess the credentials of the authors, timeliness of the research, whether or not it has been peer-reviewed, and if the content is relative to hemochromatosis and my thesis statement.

I will also be conducting a survey of healthcare professionals, WSU College of Nursing faculty and staff, and healthcare students to assess their level of knowledge about hemochromatosis. Questions will include their personal definition of hemochromatosis, how to diagnosis the disease, and what treatment looks like (please see the attached survey).

In addition, I plan to interview three generations of a family who all have HH (both homozygous and heterozygous). The interview questions will focus on the diagnosis process, the length of time diagnosis took, their individual treatment plans, and how they cope with the disease on a day-to-day basis (please see the attached list of interview questions). Before conducting the survey or interviews, I will obtain Human Studies Approval from the Office of Grants and Research Development (OGRD).

IV. Expected Results/ Potential Conclusions

Through my research, surveys, and interviews, I expect to find that the level of education and awareness about HH in the healthcare field is inadequate given the prevalence of the disease. Insufficient knowledge of HH has resulted in unnecessary complications of the disorder. These complications are not only life-threatening and often fatal, but significantly increase expenses for the individual, healthcare providers, and insurance companies.

V. Annotated Bibliography

Allen, K., Nisselle, A., Collins, V., Williamson, R., & Delatycki, M. (2008, March). Asymptomatic individuals at genetic risk of haemochromatosis take appropriate steps to prevent disease related to iron overload. *Liver International: Official Journal of the International Association for the Study of the Liver, 28*(3), 363-369. Retrieved June 19, 2009, from MEDLINE database.

This study is helpful in my research because it specifically addresses the issue of community screening for HH. Topics discussed include clinical manifestations of HH, management of the disease, long term care, and implications of community screening. The study was done over a period of 12 months and discusses how patients with HH felt about their initial diagnosis and how it impacted their lives one year later.

Bring, P., Partovi, N., Ford, J., & Yoshida, E. (2008). Iron overload disorders: treatment options for patients refractory to or intolerant of phlebotomy. *Pharmacotherapy, 28*(3), 331-342. Retrieved on June 22, 2009, from Academic Search Complete.

This article goes into extensive detail about the clinical presentation and treatment options for HH. It also includes pharmacologic options for treatment, unlike most other articles. This information will be useful in explaining side effects of HH and possible management options.

Phatak, P., Bonkovsky, H., & Kowdley, K. (2008, August 19). Hereditary hemochromatosis: Time for targeted screening. *Annals of Internal Medicine, 149*(4), 270-272. Retrieved June 19, 2009, from CINAHL with Full Text database.

This article focuses on the debate about community screening for HH. It explains different types of screening and when each type of screening should be performed. Included in the discussion is a cost analysis for both genetic and phenotypic screening.

Plaut, D., & McLellan, W. (2009). Hereditary hemochromatosis. *Journal of Continuing Education Topics & Issues, 11*(1), 18-21. Retrieved June 19, 2009, from MEDLINE database.

Plaut's article gives a general overview of HH. Topics in the paper include cause, prevalence, diagnostic tests, clinical manifestations, and treatment options of the disorder. This information will be useful to help explain the background of HH in my thesis.

Schrier, S. & Bacon, B. (2008, January 28). Clinical manifestations of hereditary hemochromatosis. Retrieved on June 22, 2009, from UpToDate database licensed to Yakima Valley Memorial Hospital.

The article by Schrier goes into great detail about the clinical manifestations of HH. It also includes information on the prevalence, different types of HH, and the pathophysiology behind the disease. Explaining the clinical manifestations of HH in my thesis will help explain why community screening and early diagnosis is essential.

Weinberg,. E.D. (2004). Exposing the hidden dangers of iron. Nashville, TN: Cumberland House Publishing, Inc.

This book explains the many different kinds of iron overload disorders, focusing on HH. It provides a thorough report of treatment, management, diagnostics, and other aspects of the disease. There are many helpful charts and graphs to further explain the disease. It also includes contact information for national voluntary health agencies and iron-related treatment products.

VI. Appendix

Survey:

What is your role in the healthcare industry?

___Physician
___Nurse Practitioner
___Pharmacist
___Nurse (RN or LPN)
___Healthcare Educator Please state healthcare field:_____
___Healthcare Student Please state healthcare field:_____
___Other: _____

Gender: Male_____ Female_____
Age:_____
Years of experience working in healthcare:_____
Have you ever heard of Hereditary Hemochromatosis? _____Yes _____No

If so, describe the condition:
_____
_____
_____

What are some common signs and symptoms of the condition?
_____
_____
_____

Name one kind of treatment for the condition:
_____
_____
_____

If you are primary care provider, how often do you diagnose Hereditary Hemochromatosis?
_____
_____
_____

If you are a primary care provider, do you ever screen for Hereditary Hemochromatosis?
_____
_____
_____

Interview Questions:

1. What year did you receive a diagnosis of Hereditary Hemochromatosis? (How long ago were you diagnosed?)
2. What symptoms (if any) did you have at the time of your diagnosis?
3. How long did you seek medical help before you received a diagnosis?
4. How many doctors/primary care providers did you see before receiving your diagnosis?
5. Do you remember your initial lab values upon diagnosis? If so, what were they?
6. How do you currently manage your disease?
7. What is your opinion about community screening for HH?
8. If you had had the option to get tested before your diagnosis, would you have done it?
9. If you have children, do you plan on getting them tested for HH or have you already had them tested?
    a. If not, why?
    b. If so, why?
10. Is there anything else you would like to share about your experience with HH?